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Letter to the Editor

Uniparental Isodisomy of Chromosome 14 in Two Cases: An Abnormal Child and a Normal Adult

To the Editor:

The Letter to the Editor by Robinson and Langlois mentions that there is an alternate possibility (mitotic recombination) to the apparent chromosome 14 isodisomy in the normal adult in our paper [Papenhausen et al., 1995] in view of the fact that only two polymorphic loci were examined. However, the likelihood of the paternal alleles being present but identical to the maternal alleles found in the index case can be estimated from the allele frequency data available for the two chromosome 14 VNTR loci that were analyzed. The approximate frequencies for random alleles at the D14S13 and D14S22 loci are 10% and 14%, respectively, and the likelihood of a second paternal haplotype matching the mother of the index case at both loci is therefore about 5%. Data from additional loci would certainly further reduce this likelihood and would be useful to examine for recombinations; however, it is much more likely that the translocation is an isochromosome even with the current data.

REFERENCE

Papenhausen PR, Mueller OT, Johnson VP, Sutcliffe M, Diamond TM, Kousseff BG (1995): Uniparental isodisomy of chromosome 14 in two cases: A dysmorphic child and a normal adult. Am J Med Genet 59:271–275.

Robinson WP, Langlois S (1996): Letter to the editor: Phenotype of maternal UPD(14). Am J Med Genet 66:89.

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